MyHospital
Department for Clinical Genetics

Molecular Genetics Report
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WHOLE EXOME SEQUENCING REPORT

Patient Name:	10000057-9	Specimen Type:	Blood, peripheral,Blood,
Date of Birth:	01/01/2000,01/01/2000,01/01/2000	Specimen Collection	02/02/2013,02/02/2013,02/02/2013
Lab Accession:	1234567,1234567,1234567	Specimen Received	01/01/2014,01/01/2014,01/01/2014
Pedigree Number:	6789000,6789000,6789000	Referring Physician:	...,...,...
Gender:	Male	Referring Facility:	Cartagenia,Cartagenia,Cartagenia
Race:	...,...,...	Referring Facility MRN:	11223344,11223344,11223344

Test performed: No target panel used
Indication for test:

TEST RESULTS

Pathogenic Variants in Genes Associated with Reported Phenotype

No variants reported.

Variants of Uncertain Significance in Genes Associated with Reported Phenoytpe

No variants reported.

Analysis Summary

Analysis summary

Nothing reported

Remarks

Authorisation

Signed off by BCM Account on 01.06.2017
Report created by BCM Account on 16.05.2017

Reference number: SCGV1-01_10000057-9

Report created: 01.06.2017

Recommendation:

Genetic testing of this individual's biological parents and other family members, particularly those who are affected, may help to clarify the significance and relative contributions of the detected variants. It is recommended that this individual and any 1st degree relative receive continued clinical evaluation and follow-up for features of DCM. Genetic counseling is recommended for this individual and their family. For assistance in locating nearby genetic counseling services please contact the laboratory at 123-456-7890. Please note that the classification of variants of unknown significance may change over time if additional information becomes available. Please contact the laboratory at 123-456-7890 once a year for any updates regarding the status of these variants.,Genetic testing of this individual's biological parents and other family members, particularly those who are affected, may help to clarify the significance and relative contributions of the detected variants. It is recommended that this individual and any 1st degree relative receive continued clinical evaluation and follow-up for features of DCM. Genetic counseling is recommended for this individual and their family. For assistance in locating nearby genetic counseling services please contact the laboratory at 123-456-7890. Please note that the classification of variants of unknown significance may change over time if additional information becomes available. Please contact the laboratory at 123-456-7890 once a year for any updates regarding the status of these variants.,Genetic testing of this individual's biological parents and other family members, particularly those who are affected, may help to clarify the significance and relative contributions of the detected variants. It is recommended that this individual and any 1st degree relative receive continued clinical evaluation and follow-up for features of DCM. Genetic counseling is recommended for this individual and their family. For assistance in locating nearby genetic counseling services please contact the laboratory at 123-456-7890. Please note that the classification of variants of unknown significance may change over time if additional information becomes available. Please contact the laboratory at 123-456-7890 once a year for any updates regarding the status of these variants.

TEST INFORMATION

Method:

Genomic DNA was extracted from the submitted specimen from this individual and submitted family members (Mother XXXXXXX, Father XXXXXXX). The Agilent SureSelect XT Human All Exon V5+UTRs kit was used to capture the exome for sequencing on an Illumina HiSeq2500 with 100bp paired-end reads. The DNA sequence was mapped to, and analyzed in comparison with, the published human genome build UCSC hg19 reference sequence. Sequencing data analysis was performed using our proprietary analytical pipeline. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for sequence changes in this individual and compared to the other provided family members.,Genomic DNA was extracted from the submitted specimen from this individual and submitted family members (Mother XXXXXXX, Father XXXXXXX). The Agilent SureSelect XT Human All Exon V5+UTRs kit was used to capture the exome for sequencing on an Illumina HiSeq2500 with 100bp paired-end reads. The DNA sequence was mapped to, and analyzed in comparison with, the published human genome build UCSC hg19 reference sequence. Sequencing data analysis was performed using our proprietary analytical pipeline. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for sequence changes in this individual and compared to the other provided family members.,Genomic DNA was extracted from the submitted specimen from this individual and submitted family members (Mother XXXXXXX, Father XXXXXXX). The Agilent SureSelect XT Human All Exon V5+UTRs kit was used to capture the exome for sequencing on an Illumina HiSeq2500 with 100bp paired-end reads. The DNA sequence was mapped to, and analyzed in comparison with, the published human genome build UCSC hg19 reference sequence. Sequencing data analysis was performed using our proprietary analytical pipeline. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for sequence changes in this individual and compared to the other provided family members.

Limitations:

...,...,...

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Disclaimer:

...,...,...

LITERATURE REFERENCES

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Annotation sources

dbSNP	Version 4	dbSNP build 147
ESP6500	Version 3	Variants in the ESP6500SI-V2 dataset of the exome sequencing project (ESP), annotated with
dbNSFP	Version 4	dbNSFP v3.0b2: Database of functional predictions for non-synonymous SNPs
1000Genomes	Version 1	1000 Genomes Phase1 release v3.20101123
1000GenomesPh	Version 1	1000 Genomes Phase 3 release v5.20130502
HGMDPublic	Version 5	HGMD® Public Database 2014.2
ClinVar	Version 6	NCBI ClinVar 20160831
COSMIC	Version 5	COSMIC release v78
ExAC	Version 1	ExAC release 0.3
OMIM	Version 1	OMIM 20160927
CIViC	Version 1	CIViC release 01-oct-2016
VariantFunction	Version 9	Transcript based Variant annotation

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